

PARENT INFORMATION for Repeat POSITIVE newborn screening results for CONGENITAL ADRENAL HYPERPLASIA (CAH)



The results of your baby's repeat screening test for congenital adrenal hyperplasia (a-DREE-nuhl HIGH-per-PLAY-zia) are presumptive positive. The initial and repeat test measured steroid 17 hydroxy progesterone (17OHP). Since the second test shows the level of 17OHP has risen, your baby should have further testing. The Nebraska Newborn Screening Program recommends referral to a pediatric endocrinologist for further diagnostic evaluation.

? Why did we have to repeat the test?

Most of the baby's that have inconclusive results on the first test will have normal results on the repeat test. Repeat testing is less expensive and is easier for most families, and is usually all that is needed.

A small percent of these babies however, will continue to have high levels of 17OHP on the repeat test. When this occurs, these babies are recommended for further evaluation by a pediatric endocrinologist who can help sort out the diagnosis. Keep in mind that new testing could show that your baby doesn't have CAH.

👉 What is the next step?

Be sure to follow your baby's doctor's instructions. She/he may recommend getting a new specimen, or refer your baby directly to a specialist. In the meantime, your doctor will talk with you about your baby's current health.

? What exactly is CAH?

CAH is caused by a missing enzyme. When this enzyme is missing, the adrenal glands produce too much of some hormones and not enough of others. The salt-retaining hormone is one that is under-produced. Therefore babies with this condition are at risk of a serious sudden "salt-wasting" episode

that can be fatal. A baby with CAH may appear normal at birth. Be sure to call your baby's doctor if any of these symptoms appear: a lot of vomiting, poor weight gain or if your baby looks ill.

There can also be other forms of CAH with less serious but still significant consequences. Treatment and monitoring is also available for these other forms of CAH. Children will usually develop normally if treatment begins in the first weeks of life. Finding out early if your baby has CAH, means you and your baby's doctor can take steps to prevent these effects.

👉 What can I do to prevent these effects?

The most important thing that you can do is to follow the recommendation of your baby's doctor. Be sure to keep any appointments made with the doctor and the specialist. If it turns out your baby does have CAH, there are very effective treatments available. A pediatric endocrinologists will be able to help you manage this through monitoring the blood levels and adjusting the medication to help normalize hormone levels.

You have probably already done this, but now is also a good time to be sure your baby is covered by your health insurance provider.

NO treatment should be done unless a physician recommends it. Therapy should NOT be started before collecting the specimen for further diagnosis.

Remember, this could be a false alarm, but if it turns out your baby does have CAH, the treatment is very effective.

For more information talk with your baby's doctor, or you can contact the Nebraska Newborn Screening Program at (402) 471-0374 for information on Pediatric Endocrinologists available in Nebraska. A good web site for more information about CAH is at www.caresfoundation.org or www.magicfoundation.org.